Prenatal diagnosis of Turner's syndrome by ultrasonography

ALASDAIR G.W. HUNTER,* MD, CM, FRCP[C]
GILBERT E. DESLAURIERS,† MD, FRCP[C], CSPQ
MARTIN S. GILLIESON,‡ MB, FRCS[C]
HENRY F. MUGGAH,§ MD, FRCS[C]

We report a case of Turner's syndrome diagnosed by ultrasonography in the second trimester of pregnancy, and discuss the findings that may be diagnostic of this condition.

Case report

A 22-year-old woman pregnant for the first time presented for uterine ultrasonography 15 weeks from the date of her last menstrual period. Fetal abnormality was not suspected, and the ultrasonography was done at the patient's request.

The initial examination revealed a fetal biparietal diameter of 29 mm, consistent with 15 weeks' gestation, a retrocervical mass and generalized edema (Fig. 1). Further examinations, at 17 and 17½ weeks' gestation, showed a septate retrocervical fluid-filled mass 5 cm in diameter (Fig. 2), massive generalized edema, ascites and polyhydramnios. The skull was normal.

The patient was advised that a normal outcome of the pregnancy was unlikely and that the diagnosis that best explained these findings was Turner's syndrome. Although she was offered amniocentesis she preferred to have the pregnancy terminated.

At the time of termination the fetus had a posterior cervical mass, typical of the cystic hygroma of Turner's syndrome, as well as generalized edema and a protruding abdomen. Autopsy revealed ascites and a single horseshoe kidney. A 45,X karyotype was confirmed by fibroblast culture and, as is frequently observed, the α-fetoprotein level in amniotic fluid drawn at the time of termination was abnormally high.[1]

Discussion

A mass in the posterior nuchal region detected by ultrasonography is likely an encephalocele or a meningocele. However, O'Brien and associates[2] detected by ultrasonography a cystic hygroma in two fetuses at 21½ and 23 weeks' gestation and emphasized that an intact cranial vault and septation in the retrocervical mass tend to rule out a neural tube defect. They did not mention fetal ascites. Although no cytogenetic studies were performed in those cases, the appearance of both fetuses after delivery was considered characteristic of Turner's syndrome.

Leonard and colleagues[3] reported the in utero detection of a probable cystic hygroma in a fetus that had already died; genetic amniocentesis revealed a 45,X karyotype. A thickened fetal outline was considered secondary to fetal death. Robinow and coworkers[4] examined a woman at 22 weeks' gestation in whom there was excessive uterine growth and found generalized fetal ascites and a retrocervical mass that was initially interpreted as an encephalocele. The pregnancy was terminated, and chromosome studies confirmed Turner's syndrome.

More recently Phillips and McGahan[5] reported several cases of cystic hygroma diagnosed by ultrasonography in the middle to late part of the second trimester. They emphasized that septation of the cervical mass and intradermal lymphangiectasia were important distinguishing features of a cystic hygroma. Edema of the abdominal wall was noted in all their cases and fetal ascites in two. They did not state whether karyotyping was done in all their cases, but two of the fetuses were reported to have a 45,X karyotype. Polyhydramnios was not present in their cases.

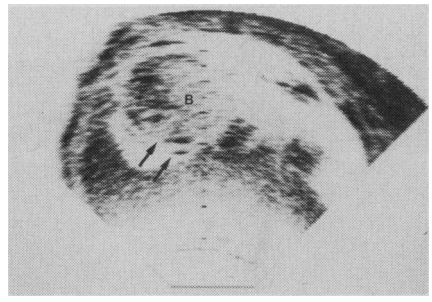

FIG. 1—Transverse ultrasound scan of fetus at 15 weeks' gestation, showing double lines (arrows) around body (B) and massive edema.

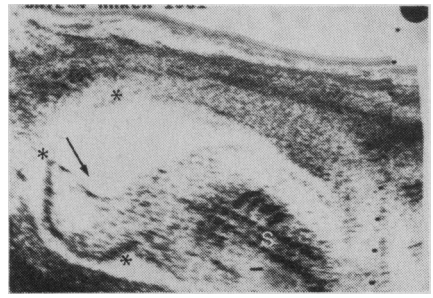

FIG. 2—Scan at 17 weeks' gestation, showing large cystic retrocervical mass (asterisks) and septum (arrow). S shows location of spine and ribs.

On the basis of our experience and that of others we believe that a posterior nuchal mass associated with generalized fetal hydrops suggests Turner's syndrome. The absence of an intracranial connection to the mass, the presence of septation within the mass, and the absence of other known causes for fetal hydrops reinforce this diagnosis.

The extensive differential diagnosis of fetal hydrops has recently been reviewed by Schwartz and associates.[6] Some of these conditions (e.g., trisomy 18) may also present the ultrasonographic appearance of cystic hygroma, but which do, and how often, is unknown. Therefore, although a normal outcome of pregnancy is unlikely under these circumstances, we believe the diagnosis should be confirmed by amniocentesis.

Turner's syndrome is more commonly diagnosed early in the second trimester than at birth because many of the fetuses with this condition die in the second trimester. The ultrasonographic findings we have described will become more frequent as the use of ultrasonography in general obstetrics and in association with genetic amniocentesis increases. Immediate appreciation of the findings should decrease the patient's period of uncertainty and avoid an unnecessary delay in diagnosis.

References

1. HUNTER A, HAMMERTON JL, BASKETT T, LYONS E: Raised amniotic-fluid alpha-fetoprotein in Turner syndrome. *Lancet* 1976; 1: 598–599
2. O'BRIEN WF, CEFALO RC, BAIR DG: Ultrasonographic diagnosis of fetal cystic hygroma. *Am J Obstet Gynecol* 1980; 138: 464–466
3. LEONARD CO, SANDERS RC, LAU HL: Prenatal diagnosis of the Turner syndrome, a familial chromosomal rearrangement and achondroplasia by amniocentesis and ultrasonography. *Johns Hopkins Med J* 1979; 145: 25–30
4. ROBINOW M, SPIRSO K, BUSCHI AJ, BRENBRIDGE ANAG: Turner syndrome: sonography showing fetal hydrops simulating hydramnios. *Am J Radiol* 1980; 135: 846–848
5. PHILLIPS HE, MCGAHAN JP: Intrauterine fetal cystic hygromas: sonographic detection. *AJR* 1981; 136: 799–802
6. SCHWARTZ SM, VISESKUL C, LADOVA R, MCPHERSON EW, GILBERT EF: Idiopathic hydrops fetalis. Report of 4 patients including 2 affected sibs. *Am J Med Genet* 1981; 8: 59–66

From *the division of genetics, Children's Hospital of Eastern Ontario, †Centre hospitalier régional de l'Outaouais, Hull, PQ, ‡the department of obstetrics and gynecology, Ottawa General Hospital and §Ottawa Civic Hospital

Reprint requests to: Dr. Alasdair G.W. Hunter, Division of genetics, Children's Hospital of Eastern Ontario, 401 Smyth Rd., Ottawa, Ont. K1H 8L1